

Stephen E Humphries

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

162 papers	19,432 citations	51 h-index	139 g-index
171 ext. papers	23,779 ext. citations	7.6 avg, IF	5.58 L-index

#	Paper	IF	Citations
162	Genetic studies of body mass index yield new insights for obesity biology. <i>Nature</i> , 2015 , 518, 197-206	50.4	2687
161	Discovery and refinement of loci associated with lipid levels. <i>Nature Genetics</i> , 2013 , 45, 1274-1283	36.3	1904
160	Familial hypercholesterolaemia is underdiagnosed and undertreated in the general population: guidance for clinicians to prevent coronary heart disease: consensus statement of the European Atherosclerosis Society. <i>European Heart Journal</i> , 2013 , 34, 3478-90a	9.5	1551
159	Defining the role of common variation in the genomic and biological architecture of adult human height. <i>Nature Genetics</i> , 2014 , 46, 1173-86	36.3	1339
158	New genetic loci link adipose and insulin biology to body fat distribution. <i>Nature</i> , 2015 , 518, 187-196	50.4	920
157	The UK10K project identifies rare variants in health and disease. <i>Nature</i> , 2015 , 526, 82-90	50.4	776
156	The interleukin-6 receptor as a target for prevention of coronary heart disease: a mendelian randomisation analysis. <i>Lancet, The</i> , 2012 , 379, 1214-24	40	658
155	Familial hypercholesterolaemia in children and adolescents: gaining decades of life by optimizing detection and treatment. <i>European Heart Journal</i> , 2015 , 36, 2425-37	9.5	430
154	A review on the diagnosis, natural history, and treatment of familial hypercholesterolaemia. <i>Atherosclerosis</i> , 2003 , 168, 1-14	3.1	426
153	Mendelian randomization of blood lipids for coronary heart disease. <i>European Heart Journal</i> , 2015 , 36, 539-50	9.5	417
152	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. <i>Diabetes</i> , 2017 , 66, 2888-2902	29.02	414
151	HMG-coenzyme A reductase inhibition, type 2 diabetes, and bodyweight: evidence from genetic analysis and randomised trials. <i>Lancet, The</i> , 2015 , 385, 351-61	40	409
150	Association between alcohol and cardiovascular disease: Mendelian randomisation analysis based on individual participant data. <i>BMJ, The</i> , 2014 , 349, g4164	5.9	406
149	Genetic causes of monogenic heterozygous familial hypercholesterolemia: a HuGE prevalence review. <i>American Journal of Epidemiology</i> , 2004 , 160, 407-20	3.8	406
148	Use of low-density lipoprotein cholesterol gene score to distinguish patients with polygenic and monogenic familial hypercholesterolaemia: a case-control study. <i>Lancet, The</i> , 2013 , 381, 1293-301	40	376
147	The polygenic nature of hypertriglyceridaemia: implications for definition, diagnosis, and management. <i>Lancet Diabetes and Endocrinology, the</i> , 2014 , 2, 655-66	18.1	357
146	Reductions in all-cause, cancer, and coronary mortality in statin-treated patients with heterozygous familial hypercholesterolaemia: a prospective registry study. <i>European Heart Journal</i> , 2008 , 29, 2625-33	9.5	321

145	Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci. <i>Nature Genetics</i> , 2015 , 47, 1415-25	36.3	292
144	Association of vitamin D status with arterial blood pressure and hypertension risk: a mendelian randomisation study. <i>Lancet Diabetes and Endocrinology,the</i> , 2014 , 2, 719-29	18.1	250
143	PCSK9 genetic variants and risk of type 2 diabetes: a mendelian randomisation study. <i>Lancet Diabetes and Endocrinology,the</i> , 2017 , 5, 97-105	18.1	225
142	Defining severe familial hypercholesterolaemia and the implications for clinical management: a consensus statement from the International Atherosclerosis Society Severe Familial Hypercholesterolemia Panel. <i>Lancet Diabetes and Endocrinology,the</i> , 2016 , 4, 850-61	18.1	215
141	Causal Associations of Adiposity and Body Fat Distribution With Coronary Heart Disease, Stroke Subtypes, and Type 2 Diabetes Mellitus: A Mendelian Randomization Analysis. <i>Circulation</i> , 2017 , 135, 2373-2388	16.7	182
140	Genetic risk factors for stroke and carotid atherosclerosis: insights into pathophysiology from candidate gene approaches. <i>Lancet Neurology, The</i> , 2004 , 3, 227-35	24.1	179
139	Association of Lipid Fractions With Risks for Coronary Artery Disease and Diabetes. <i>JAMA Cardiology</i> , 2016 , 1, 692-9	16.2	168
138	Psychological impact of genetic testing for familial hypercholesterolemia within a previously aware population: a randomized controlled trial. <i>American Journal of Medical Genetics Part A</i> , 2004 , 128A, 285-93		148
137	Cost effectiveness analysis of different approaches of screening for familial hypercholesterolaemia. <i>BMJ, The</i> , 2002 , 324, 1303	5.9	146
136	Insight into the nature of the CRP-coronary event association using Mendelian randomization. <i>International Journal of Epidemiology</i> , 2006 , 35, 922-31	7.8	137
135	Refinement of variant selection for the LDL cholesterol genetic risk score in the diagnosis of the polygenic form of clinical familial hypercholesterolemia and replication in samples from 6 countries. <i>Clinical Chemistry</i> , 2015 , 61, 231-8	5.5	130
134	Lipoprotein lipase gene variation is associated with a paternal history of premature coronary artery disease and fasting and postprandial plasma triglycerides: the European Atherosclerosis Research Study (EARS). <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 1998 , 18, 526-34	9.4	128
133	Common variants in the TCF7L2 gene and predisposition to type 2 diabetes in UK European Whites, Indian Asians and Afro-Caribbean men and women. <i>Journal of Molecular Medicine</i> , 2006 , 84, 1005-14	5.5	112
132	Development of sensitive and specific age- and gender-specific low-density lipoprotein cholesterol cutoffs for diagnosis of first-degree relatives with familial hypercholesterolaemia in cascade testing. <i>Clinical Chemistry and Laboratory Medicine</i> , 2008 , 46, 791-803	5.9	109
131	Mapping of 79 loci for 83 plasma protein biomarkers in cardiovascular disease. <i>PLoS Genetics</i> , 2017 , 13, e1006706	6	102
130	Plasma urate concentration and risk of coronary heart disease: a Mendelian randomisation analysis. <i>Lancet Diabetes and Endocrinology,the</i> , 2016 , 4, 327-36	18.1	100
129	Cross-sectional analysis of baseline data to identify the major determinants of carotid intima-media thickness in a European population: the IMPROVE study. <i>European Heart Journal</i> , 2010 , 31, 614-22	9.5	99
128	Secretory phospholipase A(2)-IIA and cardiovascular disease: a mendelian randomization study. <i>Journal of the American College of Cardiology</i> , 2013 , 62, 1966-1976	15.1	91

127	Whole exome sequencing of familial hypercholesterolaemia patients negative for LDLR/APOB/PCSK9 mutations. <i>Journal of Medical Genetics</i> , 2014 , 51, 537-44	5.8	77
126	ApoCIII gene variants modulate postprandial response to both glucose and fat tolerance tests. <i>Circulation</i> , 1999 , 99, 1872-7	16.7	77
125	Sixty-five common genetic variants and prediction of type 2 diabetes. <i>Diabetes</i> , 2015 , 64, 1830-40	0.9	76
124	Candidate gene genotypes, along with conventional risk factor assessment, improve estimation of coronary heart disease risk in healthy UK men. <i>Clinical Chemistry</i> , 2007 , 53, 8-16	5.5	74
123	Lipoprotein lipase variants D9N and N291S are associated with increased plasma triglyceride and lower high-density lipoprotein cholesterol concentrations: studies in the fasting and postprandial states: the European Atherosclerosis Research Studies. <i>Circulation</i> , 1997 , 96, 733-40	16.7	74
122	Epidemiological and genetic associations of activated factor XII concentration with factor VII activity, fibrinopeptide A concentration, and risk of coronary heart disease in men. <i>Circulation</i> , 2000 , 102, 2058-62	16.7	71
121	Improving identification of familial hypercholesterolaemia in primary care: derivation and validation of the familial hypercholesterolaemia case ascertainment tool (FAMCAT). <i>Atherosclerosis</i> , 2015 , 238, 336-43	3.1	65
120	Analysis of the frequency and spectrum of mutations recognised to cause familial hypercholesterolaemia in routine clinical practice in a UK specialist hospital lipid clinic. <i>Atherosclerosis</i> , 2013 , 229, 161-8	3.1	65
119	GWAS and colocalization analyses implicate carotid intima-media thickness and carotid plaque loci in cardiovascular outcomes. <i>Nature Communications</i> , 2018 , 9, 5141	17.4	64
118	Linkage of the cholesteryl ester transfer protein (CETP) gene to LDL particle size: use of a novel tetranucleotide repeat within the CETP promoter. <i>Circulation</i> , 2000 , 101, 2461-6	16.7	62
117	The UCL low-density lipoprotein receptor gene variant database: pathogenicity update. <i>Journal of Medical Genetics</i> , 2017 , 54, 217-223	5.8	60
116	Cost effectiveness of cascade testing for familial hypercholesterolaemia, based on data from familial hypercholesterolaemia services in the UK. <i>European Heart Journal</i> , 2017 , 38, 1832-1839	9.5	58
115	Genetic Architecture of Familial Hypercholesterolaemia. <i>Current Cardiology Reports</i> , 2017 , 19, 44	4.2	57
114	Polymorphism in the promoter region of the apolipoprotein AI gene associated with differences in apolipoprotein AI levels: the European Atherosclerosis Research Study. <i>Genetic Epidemiology</i> , 1994 , 11, 265-80	2.6	57
113	Mutational analysis in UK patients with a clinical diagnosis of familial hypercholesterolaemia: relationship with plasma lipid traits, heart disease risk and utility in relative tracing. <i>Journal of Molecular Medicine</i> , 2006 , 84, 203-14	5.5	55
112	ClinVar database of global familial hypercholesterolemia-associated DNA variants. <i>Human Mutation</i> , 2018 , 39, 1631-1640	4.7	55
111	Coronary heart disease risk prediction in the era of genome-wide association studies: current status and what the future holds. <i>Circulation</i> , 2010 , 121, 2235-48	16.7	49
110	Statin treatment of children with familial hypercholesterolemia--trying to balance incomplete evidence of long-term safety and clinical accountability: are we approaching a consensus?. <i>Atherosclerosis</i> , 2013 , 226, 315-20	3.1	48

109	Identification and management of familial hypercholesterolaemia: what does it mean to primary care?. <i>British Journal of General Practice</i> , 2009 , 59, 773-6	1.6	48
108	Greater preclinical atherosclerosis in treated monogenic familial hypercholesterolemia vs. polygenic hypercholesterolemia. <i>Atherosclerosis</i> , 2017 , 263, 405-411	3.1	47
107	A rare variant in APOC3 is associated with plasma triglyceride and VLDL levels in Europeans. <i>Nature Communications</i> , 2014 , 5, 4871	17.4	46
106	Angiotensin-I converting enzyme genotype-dependent benefit from hormone replacement therapy in isometric muscle strength and bone mineral density. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2001 , 86, 2200-4	5.6	46
105	Genetic Association of Lipids and Lipid Drug Targets With Abdominal Aortic Aneurysm: A Meta-analysis. <i>JAMA Cardiology</i> , 2018 , 3, 26-33	16.2	44
104	A genome-wide association study identifies multiple loci for variation in human ear morphology. <i>Nature Communications</i> , 2015 , 6, 7500	17.4	42
103	Carotid plaque-thickness and common carotid IMT show additive value in cardiovascular risk prediction and reclassification. <i>Atherosclerosis</i> , 2017 , 263, 412-419	3.1	39
102	Plasma Concentrations of Afamin Are Associated With Prevalent and Incident Type 2 Diabetes: A Pooled Analysis in More Than 20,000 Individuals. <i>Diabetes Care</i> , 2017 , 40, 1386-1393	14.6	39
101	Cardiovascular risk stratification in familial hypercholesterolaemia. <i>Heart</i> , 2016 , 102, 1003-8	5.1	38
100	Familial lipoprotein lipase (LPL) deficiency: a catalogue of LPL gene mutations identified in 20 patients from the UK, Sweden, and Italy. <i>Human Mutation</i> , 1997 , 10, 465-73	4.7	36
99	Association of TERC and OBFC1 haplotypes with mean leukocyte telomere length and risk for coronary heart disease. <i>PLoS ONE</i> , 2013 , 8, e83122	3.7	34
98	Population genomics of cardiometabolic traits: design of the University College London-London School of Hygiene and Tropical Medicine-Edinburgh-Bristol (UCLEB) Consortium. <i>PLoS ONE</i> , 2013 , 8, e71345	3.7	33
97	Current management of children and young people with heterozygous familial hypercholesterolaemia - HEART UK statement of care. <i>Atherosclerosis</i> , 2019 , 290, 1-8	3.1	32
96	Association of circulating metabolites with healthy diet and risk of cardiovascular disease: analysis of two cohort studies. <i>Scientific Reports</i> , 2018 , 8, 8620	4.9	32
95	Common founder mutation in the LDL receptor gene causing familial hypercholesterolaemia in the Icelandic population. <i>Human Mutation</i> , 1997 , 10, 36-44	4.7	30
94	Common genetic determinants of lung function, subclinical atherosclerosis and risk of coronary artery disease. <i>PLoS ONE</i> , 2014 , 9, e104082	3.7	30
93	The UK Paediatric Familial Hypercholesterolaemia Register: Statin-related safety and 1-year growth data. <i>Journal of Clinical Lipidology</i> , 2018 , 12, 25-32	4.9	30
92	The UK Paediatric Familial Hypercholesterolaemia Register: preliminary data. <i>Archives of Disease in Childhood</i> , 2017 , 102, 255-260	2.2	29

91	Plasma IL-5 concentration and subclinical carotid atherosclerosis. <i>Atherosclerosis</i> , 2015 , 239, 125-30	3.1	29
90	Clinical utility of the polygenic LDL-C SNP score in familial hypercholesterolemia. <i>Atherosclerosis</i> , 2018 , 277, 457-463	3.1	28
89	Statins for children with familial hypercholesterolemia. <i>The Cochrane Library</i> , 2017 , 7, CD006401	5.2	27
88	Circulating Apolipoprotein E Concentration and Cardiovascular Disease Risk: Meta-analysis of Results from Three Studies. <i>PLoS Medicine</i> , 2016 , 13, e1002146	11.6	27
87	GWAS-identified loci for coronary heart disease are associated with intima-media thickness and plaque presence at the carotid artery bulb. <i>Atherosclerosis</i> , 2015 , 239, 304-10	3.1	26
86	Sex-Specific Effects of Adiponectin on Carotid Intima-Media Thickness and Incident Cardiovascular Disease. <i>Journal of the American Heart Association</i> , 2015 , 4, e001853	6	25
85	A systematic review and meta-analysis of 130,000 individuals shows smoking does not modify the association of APOE genotype on risk of coronary heart disease. <i>Atherosclerosis</i> , 2014 , 237, 5-12	3.1	24
84	A serum 25-hydroxyvitamin D concentration-associated genetic variant in DHCR7 interacts with type 2 diabetes status to influence subclinical atherosclerosis (measured by carotid intima-media thickness). <i>Diabetologia</i> , 2014 , 57, 1159-72	10.3	24
83	The genetic architecture of the familial hyperlipidaemia syndromes: rare mutations and common variants in multiple genes. <i>Current Opinion in Lipidology</i> , 2014 , 25, 274-81	4.4	24
82	Marginal role for 53 common genetic variants in cardiovascular disease prediction. <i>Heart</i> , 2016 , 102, 1640-7	5.1	23
81	Coronary heart disease mortality in severe vs. non-severe familial hypercholesterolaemia in the Simon Broome Register. <i>Atherosclerosis</i> , 2019 , 281, 207-212	3.1	23
80	Effect of six type II diabetes susceptibility loci and an FTO variant on obesity in Pakistani subjects. <i>European Journal of Human Genetics</i> , 2016 , 24, 903-10	5.3	22
79	Universal screening at age 1-2 years as an adjunct to cascade testing for familial hypercholesterolaemia in the UK: A cost-utility analysis. <i>Atherosclerosis</i> , 2018 , 275, 434-443	3.1	22
78	Plasma autoantibodies against apolipoprotein B-100 peptide 210 in subclinical atherosclerosis. <i>Atherosclerosis</i> , 2014 , 232, 242-8	3.1	22
77	Polygenic Hypercholesterolemia and Cardiovascular Disease Risk. <i>Current Cardiology Reports</i> , 2019 , 21, 43	4.2	21
76	The genetic spectrum of familial hypercholesterolemia in south-eastern Poland. <i>Metabolism: Clinical and Experimental</i> , 2016 , 65, 48-53	12.7	20
75	Improving detection of familial hypercholesterolaemia in primary care using electronic audit and nurse-led clinics. <i>Journal of Evaluation in Clinical Practice</i> , 2016 , 22, 341-8	2.5	20
74	Low levels of IgM antibodies against phosphorylcholine are associated with fast carotid intima media thickness progression and cardiovascular risk in men. <i>Atherosclerosis</i> , 2014 , 236, 394-9	3.1	19

73	Telomere length, antioxidant status and incidence of ischaemic heart disease in type 2 diabetes. <i>International Journal of Cardiology</i> , 2016 , 216, 159-64	3.2	19
72	Statins for children with familial hypercholesterolemia. <i>The Cochrane Library</i> , 2019 , 2019,	5.2	19
71	Genetic variation in CADM2 as a link between psychological traits and obesity. <i>Scientific Reports</i> , 2019 , 9, 7339	4.9	18
70	Influence of Genetic Risk Factors on Coronary Heart Disease Occurrence in Afro-Caribbeans. <i>Canadian Journal of Cardiology</i> , 2016 , 32, 978-85	3.8	18
69	Clinical Utility of a Coronary Heart Disease Risk Prediction Gene Score in UK Healthy Middle Aged Men and in the Pakistani Population. <i>PLoS ONE</i> , 2015 , 10, e0130754	3.7	18
68	Genetic risk analysis of coronary artery disease in Pakistani subjects using a genetic risk score of 21 variants. <i>Atherosclerosis</i> , 2017 , 258, 1-7	3.1	17
67	Risk of cardiovascular disease outcomes in primary care subjects with familial hypercholesterolaemia: A cohort study. <i>Atherosclerosis</i> , 2019 , 287, 8-15	3.1	17
66	Functional analysis of four LDLR 5'UTR and promoter variants in patients with familial hypercholesterolaemia. <i>European Journal of Human Genetics</i> , 2015 , 23, 790-5	5.3	17
65	Analysis of the role of interleukin 6 receptor haplotypes in the regulation of circulating levels of inflammatory biomarkers and risk of coronary heart disease. <i>PLoS ONE</i> , 2015 , 10, e0119980	3.7	17
64	Identification of the Functional Variant(s) that Explain the Low-Density Lipoprotein Receptor (LDLR) GWAS SNP rs6511720 Association with Lower LDL-C and Risk of CHD. <i>PLoS ONE</i> , 2016 , 11, e0167676	3.7	17
63	Comparison of the characteristics at diagnosis and treatment of children with heterozygous familial hypercholesterolaemia (FH) from eight European countries. <i>Atherosclerosis</i> , 2020 , 292, 178-187	3.1	17
62	Molecular genetics of familial hypercholesterolemia in Israel-revisited. <i>Atherosclerosis</i> , 2017 , 257, 55-63	3.1	16
61	PLA2G10 Gene Variants, sPLA2 Activity, and Coronary Heart Disease Risk. <i>Circulation: Cardiovascular Genetics</i> , 2015 , 8, 356-62		15
60	The familial hypercholesterolaemia phenotype: Monogenic familial hypercholesterolaemia, polygenic hypercholesterolaemia and other causes. <i>Clinical Genetics</i> , 2020 , 97, 457-466	4	15
59	Networks in Coronary Heart Disease Genetics As a Step towards Systems Epidemiology. <i>PLoS ONE</i> , 2015 , 10, e0125876	3.7	14
58	Variants Within TSC2 Exons 25 and 31 Are Very Unlikely to Cause Clinically Diagnosable Tuberous Sclerosis. <i>Human Mutation</i> , 2016 , 37, 364-70	4.7	14
57	Common variants in the genes of triglyceride and HDL-C metabolism lack association with coronary artery disease in the Pakistani subjects. <i>Lipids in Health and Disease</i> , 2017 , 16, 24	4.4	12
56	Functional Analysis of a Carotid Intima-Media Thickness Locus Implicates BCAR1 and Suggests a Causal Variant. <i>Circulation: Cardiovascular Genetics</i> , 2015 , 8, 696-706		12

55	PCSK6 Is a Key Protease in the Control of Smooth Muscle Cell Function in Vascular Remodeling. <i>Circulation Research</i> , 2020 , 126, 571-585	15.7	12
54	Screening for familial hypercholesterolaemia in childhood: Avon Longitudinal Study of Parents and Children (ALSPAC). <i>Atherosclerosis</i> , 2017 , 260, 47-55	3.1	11
53	Genetic loci on chromosome 5 are associated with circulating levels of interleukin-5 and eosinophil count in a European population with high risk for cardiovascular disease. <i>Cytokine</i> , 2016 , 81, 1-9	4	11
52	Identifying genetic risk variants for coronary heart disease in familial hypercholesterolemia: an extreme genetics approach. <i>European Journal of Human Genetics</i> , 2015 , 23, 381-7	5.3	10
51	Post-GWAS methodologies for localisation of functional non-coding variants: ANGPTL3. <i>Atherosclerosis</i> , 2016 , 246, 193-201	3.1	10
50	The use of a highly informative CA repeat polymorphism within the abetalipoproteinaemia locus (4q22-24). <i>Prenatal Diagnosis</i> , 1997 , 17, 1181-6	3.2	10
49	Paraoxonase-1 Is Not Associated with Coronary Artery Calcification in Type 2 Diabetes: Results from the PREDICT Study. <i>Disease Markers</i> , 2012 , 33, 101-112	3.2	10
48	Influence of cytokine gene polymorphisms on proinflammatory/anti-inflammatory cytokine imbalance in premature coronary artery disease. <i>Postgraduate Medical Journal</i> , 2017 , 93, 209-214	2	9
47	Variant rs10911021 that associates with coronary heart disease in type 2 diabetes, is associated with lower concentrations of circulating HDL cholesterol and large HDL particles but not with amino acids. <i>Cardiovascular Diabetology</i> , 2016 , 15, 115	8.7	9
46	Effect of the PPARG2 Pro12Ala Polymorphism on Associations of Physical Activity and Sedentary Time with Markers of Insulin Sensitivity in Those with an Elevated Risk of Type 2 Diabetes. <i>PLoS ONE</i> , 2015 , 10, e0124062	3.7	9
45	Genome-Wide DNA Methylation in Mixed Ancestry Individuals with Diabetes and Prediabetes from South Africa. <i>International Journal of Endocrinology</i> , 2016 , 2016, 3172093	2.7	9
44	Increased Levels of Circulating Fatty Acids Are Associated with Protective Effects against Future Cardiovascular Events in Nondiabetics. <i>Journal of Proteome Research</i> , 2018 , 17, 870-878	5.6	9
43	Identifying low density lipoprotein cholesterol associated variants in the Annexin A2 (ANXA2) gene. <i>Atherosclerosis</i> , 2017 , 261, 60-68	3.1	8
42	Phenome-wide association analysis of LDL-cholesterol lowering genetic variants in PCSK9. <i>BMC Cardiovascular Disorders</i> , 2019 , 19, 240	2.3	8
41	Effectiveness of a self-management intervention with personalised genetic and lifestyle-related risk information on coronary heart disease and diabetes-related risk in type 2 diabetes (CoRDia): study protocol for a randomised controlled trial. <i>Trials</i> , 2015 , 16, 547	2.8	8
40	Human genetic evidence for involvement of CD137 in atherosclerosis. <i>Molecular Medicine</i> , 2014 , 20, 456-65	6.5	8
39	Genetics of CHD in 2016: Common and rare genetic variants and risk of CHD. <i>Nature Reviews Cardiology</i> , 2017 , 14, 73-74	14.8	7
38	Mitochondrial uncoupling proteins regulate angiotensin-converting enzyme expression: crosstalk between cellular and endocrine metabolic regulators suggested by RNA interference and genetic studies. <i>BioEssays</i> , 2016 , 38 Suppl 1, S107-18	4.1	7

37	Clinical utility gene card for: hyperlipoproteinemia, TYPE II. <i>European Journal of Human Genetics</i> , 2014 , 22,	5.3	7
36	A priori-defined Mediterranean-like dietary pattern predicts cardiovascular events better in north Europe than in Mediterranean countries. <i>International Journal of Cardiology</i> , 2019 , 282, 88-92	3.2	7
35	Cost-utility analysis of searching electronic health records and cascade testing to identify and diagnose familial hypercholesterolaemia in England and Wales. <i>Atherosclerosis</i> , 2018 , 275, 80-87	3.1	7
34	A 19-SNP coronary heart disease gene score profile in subjects with type 2 diabetes: the coronary heart disease risk in type 2 diabetes (CoRDia study) study baseline characteristics. <i>Cardiovascular Diabetology</i> , 2016 , 15, 141	8.7	6
33	How close are we to implementing a genetic risk score for coronary heart disease?. <i>Expert Review of Molecular Diagnostics</i> , 2017 , 17, 905-915	3.8	6
32	The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification.. <i>Genetics in Medicine</i> , 2021 ,	8.1	6
31	Soluble CD93 Is Involved in Metabolic Dysregulation but Does Not Influence Carotid Intima-Media Thickness. <i>Diabetes</i> , 2016 , 65, 2888-99	0.9	6
30	Effect of Coronary Artery Disease risk SNPs on serum cytokine levels and cytokine imbalance in Premature Coronary Artery Disease. <i>Cytokine</i> , 2019 , 122, 154060	4	6
29	Association of lifelong occupation and educational level with subclinical atherosclerosis in different European regions. Results from the IMPROVE study. <i>Atherosclerosis</i> , 2018 , 269, 129-137	3.1	5
28	Demonstration of the presence of the "deleted" MIR122 gene in HepG2 cells. <i>PLoS ONE</i> , 2015 , 10, e0122471	3.7	5
27	Comparison of the mutation spectrum and association with pre and post treatment lipid measures of children with heterozygous familial hypercholesterolaemia (FH) from eight European countries. <i>Atherosclerosis</i> , 2021 , 319, 108-117	3.1	5
26	Functional Analysis of the Coronary Heart Disease Risk Locus on Chromosome 21q22. <i>Disease Markers</i> , 2017 , 2017, 1096916	3.2	4
25	Higher Responsiveness to Rosuvastatin in Polygenic versus Monogenic Hypercholesterolaemia: A Propensity Score Analysis. <i>Life</i> , 2020 , 10,	3	4
24	Sex differences in cardiovascular morbidity associated with familial hypercholesterolaemia: A retrospective cohort study of the UK Simon Broome register linked to national hospital records. <i>Atherosclerosis</i> , 2020 , 315, 131-137	3.1	4
23	The overlap of genetic susceptibility to schizophrenia and cardiometabolic disease can be used to identify metabolically different groups of individuals. <i>Scientific Reports</i> , 2021 , 11, 632	4.9	4
22	Familial lipoprotein lipase (LPL) deficiency: A catalogue of LPL gene mutations identified in 20 patients from the UK, Sweden, and Italy 1997 , 10, 465		4
21	Analysis of the genetic variants associated with circulating levels of sgp130. Results from the IMPROVE study. <i>Genes and Immunity</i> , 2020 , 21, 100-108	4.4	3
20	Estimation of the prevalence of cholesteryl ester storage disorder in a cohort of patients with clinical features of familial hypercholesterolaemia. <i>Annals of Clinical Biochemistry</i> , 2019 , 56, 112-117	2.2	3

19	Applicability of the low-density lipoprotein cholesterol gene score in a South European population. <i>Atherosclerosis</i> , 2017 , 263, e99-e100	3.1	2
18	Identification of a novel proinsulin-associated SNP and demonstration that proinsulin is unlikely to be a causal factor in subclinical vascular remodelling using Mendelian randomisation. <i>Atherosclerosis</i> , 2017 , 266, 196-204	3.1	2
17	The 10-Base-Pair Insertion in the Promoter of the Factor VII Gene Is not Associated with Lower Levels of Factor VI Ic in Afrocarribeans. <i>Thrombosis and Haemostasis</i> , 1997 , 77, 213-214	7	2
16	Non-HDL or LDL cholesterol in heterozygous familial hypercholesterolaemia: findings of the Simon Broome Register. <i>Current Opinion in Lipidology</i> , 2020 , 31, 167-175	4.4	2
15	Mitochondrial uncoupling proteins regulate angiotensin-converting enzyme expression: crosstalk between cellular and endocrine metabolic regulators suggested by RNA interference and genetic studies. <i>Inside the Cell</i> , 2016 , 1, 70-81		2
14	LDL-C Concentrations and the 12-SNP LDL-C Score for Polygenic Hypercholesterolaemia in Self-Reported South Asian, Black and Caribbean Participants of the UK Biobank.. <i>Frontiers in Genetics</i> , 2022 , 13, 845498	4.5	2
13	Association of TLL1 gene polymorphism (rs1503298, T > C) with coronary heart disease in PREDICT, UDACS and ED cohorts. <i>Journal of the College of Physicians and Surgeons--Pakistan: JCPSP</i> , 2014 , 24, 615-617	8.7	2
12	Comparison of coronary heart disease genetic assessment with conventional cardiovascular risk assessment in primary care: reflections on a feasibility study. <i>Primary Health Care Research and Development</i> , 2015 , 16, 607-17	1.6	1
11	Cochrane Review: Statins for children with familial hypercholesterolemia. <i>Evidence-Based Child Health: A Cochrane Review Journal</i> , 2011 , 6, 1086-1129		1
10	Simplified detection of a mutation causing familial hypercholesterolaemia throughout Britain: evidence for an origin in a common distant ancestor. <i>Annals of Clinical Biochemistry</i> , 1998 , 35 (Pt 2), 226-35	2.2	1
9	Genetic testing for Familial Hypercholesterolaemia - Past, Present and Future. <i>Journal of Lipid Research</i> , 2021 , 100139	6.3	1
8	Management of familial hypercholesterolaemia in childhood. <i>Current Opinion in Pediatrics</i> , 2020 , 32, 633-640	3.4	1
7	The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification		1
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2	Data on the association between a simplified Mediterranean diet score and the incidence of combined, cardio and cerebro vascular events. <i>Data in Brief</i> , 2019 , 23, 103789	1.2	

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